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Delegates' Abstracts I Case presentations

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Background

Nephrogenic diabetes insipidus (NDI) is a urinary concentrating defect resulting from resistance of the renal collecting duct to the antidiuretic action of arginine vasopressin (AVP). NDI can be hereditary or acquired. The X-linked recessive form, which is the most frequent cause of inherited NDI, is caused by mutations in the gene encoding the V2 vasopressin receptor (AVPR2 gene).

Objective

To describe a Palestinian family with NDI due to a novel mutation in the AVPR2 gene.

Case Report

The proband presented during the neonatal period with failure to thrive, vomiting, irritability, fever, and polyuria of 7 ml/kg/hr. His serum sodium and osmolarity were 170 mEq/L and 330 mOsm/kg respectively with low urine osmolality of < 135 mOsm/kg. The lack of response to DDAVP and a similar picture in his older brother were in keeping with X-linked NDI. Direct sequencing the AVPR2 gene of the proband and his affected brother revealed a novel missense mutation with replacement of G by A in codon 82 of exon 2 (TGC --> TAC), predicting Cysteine to Tyrosine substitution (C82Y). The mother was a carrier for the same mutation, while the father and another healthy brother had normal AVPR2 gene.

Conclusion

To our knowledge, this is the first confirmed Palestinian family with NDI. Making the genetic diagnosis enabled us to provide a proper genetic counseling to the family with the option of future prenatal diagnosis, which would allow early diagnosis of affected males preventing severe dehydration and other NDI complications.